



# Newborn Screening Quality Assurance Program

## PROFICIENCY TESTING

## Sickle Cell Disease and Other Hemoglobinopathies

Volume 16, No. 2

Quarter 2

May 2006

### INTRODUCTION

On April 3, 2006, we distributed to all active participants the Quarter 2 proficiency testing (PT) panel consisting of five dried-blood-spot (DBS) specimens for sickle cell disease and other hemoglobinopathies. A total of 60 PT panels were mailed by overnight FedEx mail. The packages went to 52 domestic laboratories and 8 foreign laboratories. The specimen panel consisted of five DBS specimens prepared from umbilical cord blood. This PT report is a compilation of all data reports for hemoglobinopathy testing received from participants by the designated deadline date. We distribute this quarterly report to all participants, state laboratory directors, and to program colleagues by request. We received data reports from 53 newborn screening laboratories. There were 7 laboratories that did not report this quarter. All packages shipped to Brazil on April 3 were returned to CDC. ANVISA is on strike. As a result, packages can not undergo inspections and are being returned to sender. There is no word on when this will end. ❖

We requested that participants assay all survey specimens by the analytic

schemes they routinely use and report for each specimen the presumptive phenotype, the presumptive clinical assessment, and any other clinical classifications that they deem consistent with their analytic results and program operations. ❖

### PARTICIPANTS' RESULTS

The certification report listing hemoglobins (Hbs) by phenotype and their presumptive clinical assessments appears on page 2. The frequency distribution of reported phenotypes and presumptive clinical assessments appears on page 3. The individual data verification for each laboratory with evaluation comments appears on page 4. The presumptive phenotype of specimen 2635 is FA. Many laboratories detected the presence of a fast-moving additional band which is consistent with an aging or degradation band. Therefore, FAV, FAX, FAU, and FA + Fast were also acceptable results. ❖

The NSQAP will ship next quarter's PT specimens on July 10, 2006.

### SPOTLIGHT

#### Meetings

The Sickle Cell Disease Association of American (SCDAA) will conduct their 34th Annual Convention on September 27-30, 2006, at the Hyatt Regency Dallas at Reunion, Dallas, Texas. For registration information: [http://www.sicklecelldisease.org/info/2006\\_convention.phtml](http://www.sicklecelldisease.org/info/2006_convention.phtml) ❖

#### News

On March 30, 2006 James R. Eckman was publicly honored in the Senate chambers of Georgia General Assembly with a special resolution. The resolution highlighted Dr. Eckman's dedication to sickle cell patient care and newborn screening. See the following website for the entire article. [http://www.legis.ga.gov/legis/2005\\_06/search/sr1385.htm](http://www.legis.ga.gov/legis/2005_06/search/sr1385.htm). ❖

### ACKNOWLEDGMENTS

The specimens for this survey were prepared from umbilical cord blood samples supplied by Alabama State Public Health Laboratory. ❖

#### CDC/APHL

Direct inquiries to:  
Centers for Disease Control and Prevention (CDC)  
4770 Buford Highway, NE, MS/F43  
Atlanta, GA 30341-3724

This program is cosponsored by the Centers for Disease Control and Prevention (CDC) and the Association of Public Health Laboratories (APHL).

Phone : 770-488-7897  
FAX: 770-488-4255  
E-mail: NMeredith@cdc.gov

Editor : Nancy Meredith  
Production: Connie Singleton  
Sarah Brown



**Newborn Screening Quality Assurance Program  
Sickle Cell Disease and Other Hemoglobinopathies**

***Specimen and Lab Certification***

Year: 2006    Quarter: 2

**Presumptive Clinical Phenotypes**

	<b>Specimen 2631</b>	<b>Specimen 2632</b>	<b>Specimen 2633</b>	<b>Specimen 2634</b>	<b>Specimen 2635</b>
<b>Expected Presumptive Phenotype</b>	FAC	FAC	FAS	FAS	FA
<b>Accepted Presumptive Phenotypes</b>	FCA	FCA	FSA	FSA	FAV FA + fast

**Presumptive Clinical Assessments**

	<b>Specimen 2631</b>	<b>Specimen 2632</b>	<b>Specimen 2633</b>	<b>Specimen 2634</b>	<b>Specimen 2635</b>
<b>Expected Presumptive Clinical Assessment</b>	03	03	02	02	01
<b>Accepted Presumptive Clinical Assessments</b>					22

01 Normal--no abnormal Hb found  
 02 Hemoglobin S carrier  
 03 Hemoglobin C carrier  
 04 Hemoglobin SS disease (Sickle cell anemia)  
 05 Hemoglobin SC disease  
 06 Hemoglobin SD disease  
 08 Hemoglobin D carrier  
 09 Hemoglobin E carrier  
 12 Hemoglobin S, E disease

16 Alpha-thalassemia (Bart's Hb)  
 18 Hemoglobin EE disease  
 20 Assessment not listed  
 21 Unsatisfactory specimen  
 22 Unidentified variant, fast or aging band  
 NE Specimen not evaluated

**Newborn Screening Quality Assurance Program  
Sickle Cell Disease and Other Hemoglobinopathies**

**Frequency Distributions**

Year: 2006

Quarter: 2

Phenotypes			Clinical Assessments		
Specimen Number	Hemoglobin Phenotypes	Frequency Distributions	Specimen Number	Presumptive Assessments	Frequency Distributions
<b>2631</b>	FAC	50	<b>2631</b>	03 Hemoglobin C Carrier	53
	FCA	3			
<b>2632</b>	FAC	49	<b>2632</b>	03 Hemoglobin C Carrier	53
	FCA	4			
<b>2633</b>	FAS	53	<b>2633</b>	02 Hemoglobin S carrier	53
<b>2634</b>	FAS	53	<b>2634</b>	02 Hemoglobin S carrier	53
<b>2635</b>	FA	40	<b>2635</b>	01 Normal	39
	FA+FAST	4		22 Unidentified variant, fast or aging band	14
	FAU(FAST)	1			
	FAV	7			
	FAX	1			

This **NEWBORN SCREENING QUALITY ASSURANCE PROGRAM** report is an internal publication distributed to program participants and selected program colleagues. The laboratory quality assurance program is a project cosponsored by the **Centers for Disease Control and Prevention (CDC)** and the **Association of Public Health Laboratories**.

**CENTERS FOR DISEASE CONTROL AND PREVENTION (CDC)**  
**ATLANTA, GA 30341**

**Director**

Julie Louise Gerberding, M.D., M.P.H.

**Director**

**National Center for Environmental Health**

Howard Frumkin, M.D., Dr.P.H., M.P.H.

**Director**

**Division of Laboratory Sciences**

Eric J. Sampson, Ph.D.

**Chief**

**Newborn Screening Branch**

W. Harry Hannon, Ph.D.



**Contributors:** Barbara W. Adam  
Carol Bell  
Paul Dantonio  
Marie C. Earley, Ph.D.  
F. Hugh Gardner  
Sherri Hall  
L. Omar Henderson, Ph.D.  
Lisa Kalman, Ph.D.  
Lixia Li, Ph.D.  
Timothy Lim, Ph.D.  
Elizabeth McCown  
Joanne Mei, Ph.D.  
Nancy Meredith  
Nishi Patel  
Jarad Schiffer  
Anand Swamy, Ph.D.  
Robert Vogt, Ph.D.  
Yingtao Zhou

**Production:** Sarah Brown  
Felicia Manning  
Connie Singleton

**ASSOCIATION OF PUBLIC HEALTH LABORATORIES**  
**WASHINGTON, DC 20036-3320**

**President**

Katherine Kelley, Dr.P.H.

**Chairman, Newborn Screening and Genetics in Public Health Committee**

William Becker, D.O., M.P.H.

**Chairman, Newborn Screening Quality Assurance Subcommittee**

John Sherwin, Ph.D.



**INQUIRIES TO:**

Nancy Meredith, Editor • Centers for Disease Control and Prevention (CDC)  
Newborn Screening Quality Assurance Program • Mailstop F-43  
4770 Buford Highway, N.E. • Atlanta, GA 30341-3724  
Phone (770) 488-4582 • FAX (770) 488-4255 • E-mail: NMeredith@cdc.gov